REPORTS OF SIX CASES OF FRIEDREICH'S ATAXIA, OCCURRING IN THREE DIFFERENT FAMILIES.

By DR. CHARLES W. ROOK, of Quincy, Ill., and DR. CHARLES L. DANA, of New York.

THE histories of the first four cases were sent to me, with photographs, by Dr. Rook. The histories given leave no doubt in my mind that they are true cases of Friedreich's ataxia.

To them I have added one case, recently observed by me, and a second, which was sent to me by Dr. J. W. S. Gouley, and which has been under my observation for several years. This latter patient's history was reported in brief in the "Medical Record," of October 1, 1887. It will appear soon, in full, in an article on Friedreich's ataxia, by myself, in Keating's "Cyclopædia of Children's Diseases." I therefore omit the publication of the case here. Some comments upon the disease, and some new observations made upon my cases, have been embodied in the article referred to, and cannot properly be published now.

I find that there have now been reported about 165 cases. Fifty-four, or one-third of them, have been reported by seventeen different American observers. I conclude that Friedreich's Ataxia is a relatively frequent disease in this country.

C. L. D.

HISTORY OF DR. DANA'S CASES.

CASE I.—B. N., United States, aged twelve. One paternal uncle had some spinal trouble; one paternal aunt had some spinal trouble, and wore braces from tenth to forty-fifth year; father had some spinal trouble, and walked ataxic in last ten years of life, when he died of pneumonia. Patient is youngest of four children: one boy, twenty-four; one boy, twenty; one girl, fifteen—all of whom are well.

Birth natural, at full term. He seemed well until his seventh year, when he had scarlet fever. Has been in poorer health since, but no especial trouble was noted until the present one developed. No history of diphtheria. He had whooping-cough in the spring of 1888. In the following winter of 1888–'89 he was noticed to stumble in his gait while playing, and especially to stumble in the dark. His gait was awkward and irregular. He complained of no pain in the legs, but had some pains in head and stomach occasionally. No bladder or rectal troubles, and no disorders of vision or speech were complained of.

He was sent to Dr. S. N. Phelps, hip-disease being suspected. Dr. Phelps excluded any such trouble, and referred him to me, October 16, 1889.

When examined by me he showed a gait somewhat ataxic and stumbling, but also with an apparent limp in the right leg. He could not walk far with the eyes closed, nor could he stand with the eyes closed and the feet together except for a short time. He was well grown, the legs being a little small, but not showing any local atrophy or hypertrophy. His speech was somewhat peculiar; but this it had always been. No nystagmus; vision and optic nerves normal. His knee-jerks were present. Slight bactile anæsthesia over right foot, and dorsal flexion of right great toe were noted. Urine normal; electrical reactions normal.

He had no pain; no spinal curvature. Essentially his trouble was only an ataxic gait; the arms not being affected. This ataxia was very apparent, however, and was worse in the dark; so that though clumsy in gait at all times, he was especially so at night.

The case is in its incipient stage, and all the symptoms are not present; but, in view of the dominance of ataxia and the family history, there seems to be no doubt of the correctness of the diagnosis.

CASE IV.—J. D., aged twenty-one, United States. No family history of ataxia or other nervous disease. It developed at the age of fifteen, after a blow on the head, and was associated with polyuria and speech-disturbances. The ataxia and the peculiar rolling gait are very marked.

HISTORY OF DR. ROOK'S CASES.

Their maternal grandfather died of phthisis at the age of seventy years. Their grandmother is sixty-eight years old, and enjoys good health. To them were born eight children. One son died in convulsions when nine months

old, and another of phthisis at the age of twenty-two years. Three sons, in good health, with families in which no ataxia has developed. The three daughters have good health. One is single; one is married seven years, but has no children; and one, the mother of my cases. She is of medium build, and forty-three years old. She has had no serious sickness. Intoxicants, chiefly beer, are employed as a beverage in the family of the latter.

In the family history there are no instances of consanguinity, syphilis, insanity, or ataxia. The only predisposing causes revealed are phthisis and alcoholism.

To Mr. and Mrs. G., parents of my cases, were born eight children, as follows:

1st. Julia, aged twenty years, ataxic since the age of eleven years.

2d. Laurence, aged eighteen years, not ataxic.

3d. Antone, aged sixteen years, ataxic since the age of eleven years.

4th. Clara, aged fourteen years, ataxic since the age of eleven years.

5th. Katie, aged twelve years, not ataxic.

6th. Bertha, aged nine years, ataxia developing.

7th. Infant daughter, died of whooping-cough at the age of sixteen months.

8th. Infant daughter, died of inanition at the age of seventeen months.

CASE I.—Julia G., aged twenty years. Her early life was very free of sickness. In her second year she had some fever during dentition; and in her eleventh year an attack of measles, from which she recovered without complications.

During her eleventh year she first experienced a sense of weakness in the lower limbs, and in a few months her gait became staggering. These symptoms gradually increased till she was unable to walk or stand without support. Her arms were affected one year later, or during her twelfth year. Since the commencement of this disease she has had no other sickness.

She is now much deformed by contractures. There exists a kyphosis, a left dorsal and a right lumbar scoliosis, a double talipes valgus; when at rest, the hands assume a wrist-drop and the fingers a claw-like appearance.

She is not well nourished, though the food taken is digested without distress. Physical examination of lungs and heart negative. Respiration 18 and the pulse 80 per minute. Urine, reaction acid, sp. gr. 1.020, and contained no morbid elements.

The mammæ are partially developed, and the menses have thrice occurred, at intervals of two or three months, during this her twentieth year.

The expressionless appearance of her face is less marked when attentively listening to or engaging in conversation. When reading she pronounces slowly yet distinctly, but in conversation her speech is slower, voice more tremulous, and acts of cerebration are performed with an effort. She has a good memory, as shown by the fact that she can yet instruct her brothers and sisters in reading and mathematics, though it is eight years since she was at school.

Her sleep is natural in appearance, though she requires from nine to fourteen hours daily. Her disposition is becoming more irritable than in the earlier years of her affliction.

Decided atrophic changes have occurred throughout the entire muscular system. No muscles are paralyzed, though their electrical reaction is much less than normal.

Co-ordination is very deficient, more so in the lower than upper extremities. Though unable to stand or walk without support, ataxia of station is probably increased by closure of her eyes, for then the movements of her arms become more unsteady.

She is able to feed and dress herself and perform some work, as the washing and drying of dishes and sewing. Several minutes are required for her to thread her needle, and then only by supporting one hand with the other.

She has some attacks of vertigo, usually soon after rising of a morning or after meals. She often experiences a feeling of numbness in her limbs, but no pain.

Cutaneous sensibility is diminished, particularly in the lower limbs, where two points of pressure may be separated as much as four inches and described as only one point of pressure.

Her vision is good, no nystagmus, and ophthalmoscopic examination of the eyes negative.

The sense of hearing, taste, and smell normal. Superficial reflexes, as the plantar, are diminished. Ankle clonus and patellar reflexes absent.

The extremities are colder than natural, but not œdematous. No secretory disturbances, save that of the menses.

CASE II.—Antone G., aged sixteen years. When seven years old he had measles, from which he recovered without complications. During his eleventh year his parents first noticed his staggering gait, and about one year later his arms became affected.

There now exist no contractures except a marked kyphosis, which can be partially overcome at will, and a double talipes valgus.

His gait is very ataxic, the feet being separated some six or eight inches in order to better maintain his equilibrium while standing or walking.

Over slight obstructions he stumbles, occasionally falling; yet, notwithstanding this difficulty, he has, during this year, taken many walks of two or three miles.

His nutrition is good, and the functions of the primæ viæ are normal.

Physical examination of lungs and heart negative, though he has frequent attacks of palpitation.

On three occasions were witnessed attacks of syncope that are worthy of record. The first attack occurred while in the Sayre suspension-apparatus; second, while standing and being examined for spinal curvature; and third, while sitting and being examined to determine the electrical reaction of the muscles.

The strength of current employed, that induced the last attack, was only sufficient to produce, with the electrodes placed on the right arm, at elbow and wrist, very slight flexion of the hand.

He would announce the onset of the attacks by the remark, "I feel so dizzy." His head would sway from side to side, face become pale, radial pulse disappear, muscles relax, and unconsciousness supervene.

The relaxed condition would continue for possibly half a minute, during which time the pulse and respiration were not perceptible; then followed a sudden and violent tonic convulsion, his body assuming a position of extreme opisthotnos. In about one minute the convulsive state began to relax, consciousness returned, and for some minutes he suffered severe pain in the erector spinæ muscles. During the seizure the pulse and respiration returned, the latter being stertorous, and the kyphosis completely disappeared.

Temperature is normal, respiration 19, and pulse 84.

The urine is acid in reaction, specific gravity 1.025, and contained no morbid elements.

His intellect is but little affected, memory good, speech slow, voice tremulous, and facial expression habitually gloomy.

His sleep is natural, save when occasionally disturbed by spasmodic contractions of the lower limbs. These contractions are painless.

No paralysis or atrophy exists. Electrical reaction of muscles diminished.

His power of co-ordination is much less than normal; yet, under direct supervision of sight, he has fair control of his extremities. With eyes closed, the ataxia is more apparent, for then he can neither stand nor walk without support.

He has frequent attacks of vertigo, always preceding attacks of syncope. He experiences no pain, save after the convulsive seizures, but often has a feeling of numbness in the extremities.

Cutaneous sensibility less than normal; more noticeable in lower extremities, where he cannot distinguish between two points of pressure, if separated three or four inches; nor can he correctly locate a point of pressure.

Vision is good, and ophthalmoscopic examination of the eyes negative.

Sense of hearing, taste, and smell normal.

Plantar reflex diminished. Ankle clonus and patellar reflex absent.

The only secretory disturbance has been an occasional incontinence of urine.

There are no vaso-motor changes.

CASE III.—Clara G., aged fourteen years. When two years old she had an attack of pneumonia, and has since remained delicate. At five years of age she had measles, and recovered without complications.

During her eleventh year the ataxia began in the lower limbs, and within a year the arms were also affected.

The first objective symptom noticed was her staggering gait; she also early experienced a sense of weakness in the limbs.

Her gait is now very ataxic, as are also the movements of her arms. In passing through a room she will touch one or more pieces of furniture, thereby enabling her to better maintain her equilibrium.

There exist some contractures, as a kyphosis, double talipes valgus, and wrist-drop, all of which can be partially overcome.

She has a dry, hacking cough, and auscultation reveals the presence of numerous crepitant rales in each lung, but more abundant in the posterior part of the lower lobes.

The heart's action is very irritable, hastily crossing a room causing palpitation for several minutes.

Her temperature, as shown by a thermometer, is usually above normal, though the extremities feel colder than natural. There is frequent flushing of one or both cheeks. This increase of temperature and hectic is due to the diseased condition of the lungs.

Respiration 21, pulse 90.

Urine is acid, with a specific gravity of 1.024, and in other respects normal.

Her facial expression is more intelligent than her sister Julia's or brother Antone's, for it is not melancholic.

She learns easily, has a good memory; speech slow and voice very tremulous.

Sleep is natural.

Co-ordination very much diminished. She cannot walk in a straight line with eyes open, or stand with feet together without her body swaying, and will fall, if not supported, when her eyes are closed.

The ataxia is nearly as marked in the upper as in the lower extremities. When extending the hand to grasp an object, its claw-like appearance is quite noticeable.

Electrical reaction of muscles diminished. Atrophic changes are marked throughout the muscular system.

No muscle or group of muscles is paralyzed.

She has frequent attacks of vertigo, and is occasionally disturbed by a feeling of numbness in the lower limbs, but has no pain.

Cutaneous sensibility is diminished. Not able to locate or distinguish between two points of pressure any better than her sister or brother.

Ophthalmoscopic examination of her eyes negative. Vision is good. Some months ago nystagmus was quite marked; now it is only occasionally observed.

Her sense of hearing, taste, and smell normal.

Plantar reflex diminished. Ankle clonus and patellar reflex are absent.

There are no secretory disturbances.

CASE IV.—Bertha G., aged nine years. At the time of her birth the other children of the family were sick with the measles, and nine days later a cough developed and a slight eruption appeared.

She was supposed to have had the measles, When one and a half years old she had convulsions, and once or twice yearly the attacks recurred till she was six years old. The

later attacks are known to be due to indigestion, and probably the earlier ones also.

When she was seven years of age she had the measles, there being at that time an epidemic of this disease; but no other member of the family was attacked.

A slight deafness was observed after her recovery; and two months later there suddenly appeared, at each ear, a profuse otorrhœa, which ceased in a few weeks, leaving her almost entirely deaf.

She is now in good health, there being no derangement of the digestion, heart, lungs, or kidneys.

Her disposition is pleasant, and her face bright and intelligent. Speech is slow, but not tremulous. Sleep is natural.

Ataxia of locomotion or station is not apparent with eyes open, but with them closed her gait and station are each unsteady.

In the excitement of play, or when eyes are closed, the motion of her arms is also slightly ataxic.

There are no muscles atrophied, contractured, or paralyzed. Electrical reaction of muscles normal. Tactile sensation diminished. Vision, taste, and smell normal, but hearing destroyed.

Plantar reflex nearly normal. Ankle clonus absent, and patellar reflex present, but greatly diminished.

There are no vaso-motor or secretory disturbances.

The treatment employed for these patients has been, for each: silver nitrate, one grain, in pill, twice daily; for Cases I. and III., cod-liver oil in emulsion, and, for all, suspension. For six months an average of two suspensions per week have been given.

Results of Treatment.—The general health of Cases I. and III. are improved, but no change noted in their ataxia. Case II. is improved in his gait, but not in his reflexes or tactile sense. Case IV.: Ataxia not increased during six months' treatment.